

AperTO - Archivio Istituzionale Open Access dell'Università di Torino

Novel insights into the relationship between KRIT1 and ROS homeostasis: KRIT1 loss-of-function causes a ROS-dependent upregulation of transcription factors involved in oxidative stress response.

This is the author's manuscript

Original Citation:

Availability:

This version is available <http://hdl.handle.net/2318/124033> since

Publisher:

Angioma Alliance

Terms of use:

Open Access

Anyone can freely access the full text of works made available as "Open Access". Works made available under a Creative Commons license can be used according to the terms and conditions of said license. Use of all other works requires consent of the right holder (author or publisher) if not exempted from copyright protection by the applicable law.

(Article begins on next page)

8th Annual Angioma Alliance
CCM SCIENTIFIC MEETING

November 15-16, 2012

DoubleTree Bethesda – Washington, DC
Ballroom D

DAY 1 | THURSDAY, NOVEMBER 15TH

7:45 **WELCOME & OPENING REMARKS**
Amy Akers & Sara Sukalich – Angioma Alliance

SESSION I – PROTEOMICS, STRUCTURE & FUNCTION
DOUG MARCHUK, SESSION CHAIR

8:00 ***New insights into the structure and function of CCM proteins***
Titus Boggon – Yale University School of Medicine

8:20 ***Proteomics characterization of CCM complexes***
Anne-Claude Gingras – Samuel Lunenfeld Research Institute

8:40 ***Structural Basis of the Junctional Anchorage of the Cerebral
Cavernous Malformations Complex***
Alexandre Gingras – University of California San Diego

SESSION II – SIGNALING
BRENT DERRY, SESSION CHAIR

9:00 ***Defining the Ccm3 signaling pathway in a zebrafish model of CCM
disease***
Bilge Yoruk** – Sick Kids Research Institute

9:20 ***CCM3 and senescence***
Juan Zalvide – University of Santiago de Compostela

9:40 *COFFEE BREAK*

10:00 ***CCM3 functions in brain development***
Angeliki Louvi – Yale School of Medicine

10:20 ***CCM3 regulates endosome recycling in the C. elegans excretory cell***
Ben Lant – Sick Kids Research Institute

- 10:40 ***Novel Endothelial Signaling in CCM***
Rebecca Stockton – University of California Los Angeles
- 11:00 ***Further Studies of Fasudil Treatment in Murine Models of Cerebral Cavernous Malformation Disease***
Robert Shenkar – University of Chicago
- 11:20 DISCUSSION OF SESSIONS I & II
- 12:00 Lunch | Oz Restaurant

SESSION III – VASCULAR BIOLOGY & INFLAMMATION

BRANT WEINSTEIN, SESSION CHAIR

- 1:00 ***The CCM2 paralogue CCM2L opposes canonical cerebral cavernous malformation signaling in endothelial cells during cardiovascular growth***
Xiangjian Zheng – University of Pennsylvania
- 1:20 ***Loss of Notch signaling in the adult endothelium: implications for CCM***
Andreas Fischer – German Cancer Research Center Heidelberg (DKFZ)
- 1:40 ***The recombinant antibody construction and restricted B cell repertoire in Human Cerebral Cavernous Malformation (CCM)***
Changbin Shi – University of Chicago
- 2:00 ***Decreased KRIT1 expression leads to increased vascular permeability and modifies inflammatory responses in vivo.***
Angela Glading – University of Rochester
- 2:20 ***CCM2 intersects a novel pathway of cytokine mediated vascular instability***
Dean Li – University of Utah
- 2:40 *COFFEE BREAK*

SESSION IV – LESION GENESIS

KEVIN WHITEHEAD, SESSION CHAIR

- 3:00 ***Novel insights into the relationship between KRIT1 and ROS homeostasis: KRIT1 loss-of-function causes a ROS-dependent upregulation of transcription factors involved in oxidative stress response***
Saverio Francesco Retta – University of Torino

- 3:20 ***Exploring the Implications of a Two-Hit Mechanism in Cerebral Cavernous Malformations***
David McDonald** - Duke University Medical Center
- 3:40 ***CCM3-dependent EC-SMC/pericyte interactions in CCM lesion development mouse models and mechanistic studies***
Wang Min – Yale University
- 4:00 ***Angiogenesis is Required for Cavernous Malformation Development***
Kevin Whitehead – University of Utah
- 4:20 *DISCUSSION OF SESSIONS III & IV*
- 5:00 *END OF DAY 1*
- 7:00 DINNER | BALLROOM C

DAY 2 | Friday, November 16th

- 8:30 **WELCOME**
Connie Lee – Angioma Alliance & CCM3 Action

SESSION V – MAGNETIC RESONANCE IMAGING TECHNOLOGIES

LESLIE MORRISON, SESSION CHAIR

- 8:40 ***Quantitative Iron Burden as a Biomarker of Cumulative Hemorrhages in Cerebral Cavernous Malformations: Studies in Mouse and Man***
Luying (Ryan) Li** – West China Medical School of Sichuan University & University of Chicago
- 9:00 ***Novel Magnetic Resonance Imaging Biomarkers of Human CCM Disease: Dynamic Contrast-Enhanced Quantitative Perfusion***
Abdul Ghani Mikati** – University of Chicago
- 9:20 ***White Matter Hyperintensities in CHM CCM1***
Blaine Hart – University of New Mexico
- 9:40 *COFFEE BREAK*

SESSION VI – CLINICAL STUDIES

ISSAM AWAD, SESSION CHAIR

- 10:00 ***Spectrum of Human Causative Mutations in the KRIT1, CCM2 and PDCD10 Genes***
James Weber – PreventionGenetics

- 10:20 ***Clinical Factors Associated with Lesion Count in Familial Cerebral Cavernous Malformation Type 1 Patients with the Common Hispanic Mutation***
Hélène Choquet** – University of California San Francisco
- 10:40 ***Cutaneous Features of the CCM1-CHM Cohort***
Leslie Morrison – University of New Mexico
- 11:00 ***Outcome after surgical or conservative management of cerebral cavernous malformations: a prospective, population-based cohort study***
Margaret A. Horne** - University of Edinburgh
- 11:20 DISCUSSION OF SESSION V & VI
- 12:00 LUNCH | OZ RESTAURANT

Session VII – Panel Discussion of Clinical Trails for CCM

- 1:00 ***Biomarkers***
Issam Awad – University of Chicago
- 1:10 ***Recruitment Strategies***
Leslie Morrison – University of New Mexico
- 1:20 ***Trials & Research Consortia***
William Young – University of California San Francisco
- 1:30 ***Food & Drug Administration Perspective***
Gumei Liu – Rare Diseases Program Office of New Drugs
- 1:40 ***National Institutes of Health Perspective***
Claudia Moy – NINDS office of Clinical Research
- 1:50 OPEN DISCUSSION
- 3:00 CLOSE OF MEETING

MEETING SPONSORS

ANGIOMA ALLIANCE
NATIONAL INSTITUTE OF NEUROLOGICAL DISORDERS & STROKE
CCM3 ACTION
INNOLYST (PATIENT CROSSROADS)

**Trainee Travel Award Winner